carnitine palmitoyltransferase I deficiency

Carnitine palmitoyltransferase I (CPT I) deficiency is a condition that prevents the body from using certain fats for energy, particularly during periods without food (fasting). The severity of this condition varies among affected individuals.

Signs and symptoms of CPT I deficiency often appear during early childhood. Affected individuals usually have low blood sugar (hypoglycemia) and a low level of ketones, which are produced during the breakdown of fats and used for energy. Together these signs are called hypoketotic hypoglycemia. People with CPT I deficiency can also have an enlarged liver (hepatomegaly), liver malfunction, and elevated levels of carnitine in the blood. Carnitine, a natural substance acquired mostly through the diet, is used by cells to process fats and produce energy. Individuals with CPT I deficiency are at risk for nervous system damage, liver failure, seizures, coma, and sudden death.

Problems related to CPT I deficiency can be triggered by periods of fasting or by illnesses such as viral infections. This disorder is sometimes mistaken for Reye syndrome, a severe disorder that may develop in children while they appear to be recovering from viral infections such as chicken pox or flu. Most cases of Reye syndrome are associated with the use of aspirin during these viral infections.

Frequency

CPT I deficiency is a rare disorder; fewer than 50 affected individuals have been identified. This disorder may be more common in the Hutterite and Inuit populations.

Genetic Changes

Mutations in the *CPT1A* gene cause CPT I deficiency. This gene provides instructions for making an enzyme called carnitine palmitoyltransferase 1A, which is found in the liver. Carnitine palmitoyltransferase 1A is essential for fatty acid oxidation, which is the multistep process that breaks down (metabolizes) fats and converts them into energy. Fatty acid oxidation takes place within mitochondria, which are the energy-producing centers in cells. A group of fats called long-chain fatty acids cannot enter mitochondria unless they are attached to carnitine. Carnitine palmitoyltransferase 1A connects carnitine to long-chain fatty acids so they can enter mitochondria and be used to produce energy. During periods of fasting, long-chain fatty acids are an important energy source for the liver and other tissues.

Mutations in the *CPT1A* gene severely reduce or eliminate the activity of carnitine palmitoyltransferase 1A. Without enough of this enzyme, carnitine is not attached to long-chain fatty acids. As a result, these fatty acids cannot enter mitochondria and be converted into energy. Reduced energy production can lead to some of the features of

CPT I deficiency, such as hypoketotic hypoglycemia. Fatty acids may also build up in cells and damage the liver, heart, and brain. This abnormal buildup causes the other signs and symptoms of the disorder.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- carnitine palmitoyltransferase IA deficiency
- CPT 1A deficiency
- CPT deficiency, hepatic, type I
- CPT I deficiency
- liver form of carnitine palmitoyltransferase deficiency

Diagnosis & Management

These resources address the diagnosis or management of CPT I deficiency:

- Baby's First Test
  http://www.babysfirsttest.org/newborn-screening/conditions/carnitine-palmitoyltransferase-i-deficiency
- FOD (Fatty Oxidation Disorders) Family Support Group: Diagnostic Approach to Disorders of Fat Oxidation - Information for Clinicians
  http://www.fodsupport.org/clinicians.htm
- GeneReview: Carnitine Palmitoyltransferase 1A Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1527
- Genetic Testing Registry: Carnitine palmitoyltransferase I deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342789/

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Health Topic: Lipid Metabolism Disorders
  https://medlineplus.gov/lipidmetabolismdisorders.html
- Health Topic: Mitochondrial Diseases
  https://medlineplus.gov/mitochondrialdiseases.html
- Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

Genetic and Rare Diseases Information Center

- Carnitine palmitoyl transferase 1 deficiency
  https://rarediseases.info.nih.gov/diseases/1120/carnitine-palmitoyl-transferase-1-deficiency

Educational Resources

- Children Living with Inherited Metabolic Diseases (CLIMB): Carnitine Palmitoyltransferase I Deficiency Fact Sheet
  http://www.climb.org.uk/IMD/Charlie/CarnitinePalmitoylTransferaseDeficiencyType%201.pdf
- MalaCards: carnitine palmitoyltransferase i deficiency , muscle
  http://www.malacards.org/card/carnitine_palmitoyltransferase_i_deficiency_muscle
- New England Consortium of Metabolic Programs
  http://newenglandconsortium.org/for-families/other-metabolic-disorders/fatty-acid-oxidation-disorders/cpt-i-deficiency/
- Orphanet: Carnitine palmitoyl transferase 1A deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=156
- Screening, Technology, and Research in Genetics
  http://www.newbornscreening.info/Parents/fattyaciddisorders/CPT1.html

Patient Support and Advocacy Resources

- Children Living with Inherited Metabolic Diseases (CLIMB)
  http://www.climb.org.uk/
- FOD (Fatty Oxidation Disorders) Family Support Group
  http://www.fodsupport.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/carnitine-palmitoyltransferase-1a-deficiency/
- United Mitochondrial Disease Foundation
  http://www.umdf.org/

GeneReviews

- Carnitine Palmitoyltransferase 1A Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1527

Genetic Testing Registry

- Carnitine palmitoyltransferase I deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342789/

ACT Sheets

- Elevated C0/C16+C18
  https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/C0_C16_C18.pdf

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22carnitine+palmitoyltransferase+I+deficiency%22

Scientific articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28carnitine+palmitoyltransferase+1a+deficiency%5BALL%5D%29+OR+%28carnitine+palmitoyltransferase+1+deficiency%5BALL%5D%29+OR+%28CPT1A+deficiency%5BALL%5D%29+OR+%28carnitine+palmitoyltransferase+type+1a%29%29+AND+human%5Bmh%5D

OMIM

- CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY
  http://omim.org/entry/255120

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